



incontinentia pigmenti

Incontinentia pigmenti is a condition that can affect many body systems, particularly the skin. This condition occurs much more often in females than in males.

Incontinentia pigmenti is characterized by skin abnormalities that evolve throughout childhood and young adulthood. Many affected infants have a blistering rash at birth and in early infancy, which heals and is followed by the development of wart-like skin growths. In early childhood, the skin develops grey or brown patches (hyperpigmentation) that occur in a swirled pattern. These patches fade with time, and adults with incontinentia pigmenti usually have lines of unusually light-colored skin (hypopigmentation) on their arms and legs.

Other signs and symptoms of incontinentia pigmenti can include hair loss (alopecia) affecting the scalp and other parts of the body, dental abnormalities (such as small teeth or few teeth), eye abnormalities that can lead to vision loss, and lined or pitted fingernails and toenails. Most people with incontinentia pigmenti have normal intelligence; however, this condition may affect the brain. Associated problems can include delayed development or intellectual disability, seizures, and other neurological problems.

Frequency

Incontinentia pigmenti is an uncommon disorder. Between 900 and 1,200 affected individuals have been reported in the scientific literature. Most of these individuals are female, but several dozen males with incontinentia pigmenti have also been identified.

Genetic Changes

Mutations in the *IKBKG* gene cause incontinentia pigmenti. The *IKBKG* gene provides instructions for making a protein that helps regulate nuclear factor-kappa-B. Nuclear factor-kappa-B is a group of related proteins that helps protect cells from self-destructing (undergoing apoptosis) in response to certain signals.

About 80 percent of affected individuals have a mutation that deletes some genetic material from the *IKBKG* gene. This deletion probably leads to the production of an abnormally small, nonfunctional version of the IKBKG protein. Other people with incontinentia pigmenti have mutations that prevent the production of any IKBKG protein. Without this protein, nuclear factor-kappa-B is not regulated properly, and cells are more sensitive to signals that trigger them to self-destruct. Researchers believe that this abnormal cell death leads to the signs and symptoms of incontinentia pigmenti.

Inheritance Pattern

This condition is inherited in an X-linked dominant pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In females (who have two X chromosomes), a mutation in one of the two copies of the gene in each cell is sufficient to cause the disorder. Some cells produce a normal amount of *IKBKG* protein and other cells produce none. The resulting imbalance in cells producing this protein leads to the signs and symptoms of incontinentia pigmenti.

In males (who have only one X chromosome), most *IKBKG* mutations result in a total loss of the *IKBKG* protein. A lack of this protein appears to be lethal early in development, so few males are born with incontinentia pigmenti. Affected males who survive may have an *IKBKG* mutation with relatively mild effects, an *IKBKG* mutation in only some of the body's cells (mosaicism), or an extra copy of the X chromosome in each cell.

Some people with incontinentia pigmenti inherit an *IKBKG* mutation from one affected parent. Other cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

Other Names for This Condition

- Bloch-Siemens-Sulzberger Syndrome
- Bloch-Siemens syndrome
- Bloch-Sulzberger Syndrome
- IP

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Incontinentia pigmenti syndrome
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0021171/>

Other Diagnosis and Management Resources

- GeneReview: Incontinentia Pigmenti
<https://www.ncbi.nlm.nih.gov/books/NBK1472>
- MedlinePlus Encyclopedia: Incontinentia Pigmenti Syndrome
<https://medlineplus.gov/ency/article/001583.htm>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Incontinentia Pigmenti Syndrome
<https://medlineplus.gov/ency/article/001583.htm>
- Health Topic: Skin Conditions
<https://medlineplus.gov/skinconditions.html>
- Health Topic: Skin Pigmentation Disorders
<https://medlineplus.gov/skinpigmentationdisorders.html>

Genetic and Rare Diseases Information Center

- Incontinentia pigmenti
<https://rarediseases.info.nih.gov/diseases/6778/incontinentia-pigmenti>

Additional NIH Resources

- National Institute of Neurological Disorders and Stroke
<https://www.ninds.nih.gov/Disorders/All-Disorders/Incontinentia-pigmenti-Information-Page>

Educational Resources

- Disease InfoSearch: Incontinentia pigmenti
<http://www.diseaseinfosearch.org/Incontinentia+pigmenti/3798>
- MalaCards: incontinentia pigmenti
http://www.malacards.org/card/incontinentia_pigmenti
- Orphanet: Incontinentia pigmenti
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=464

Patient Support and Advocacy Resources

- Incontinentia Pigmenti International Foundation
<http://www.ipif.org>
- National Foundation for Ectodermal Dysplasias
<https://www.nfed.org/learn/types/>
- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/incontinentia-pigmenti/>
- Resource list from the University of Kansas Medical Center
<http://www.kumc.edu/gec/support/incontin.html>

GeneReviews

- Incontinentia Pigmenti
<https://www.ncbi.nlm.nih.gov/books/NBK1472>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22incontinentia+pigmenti%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Incontinentia+Pigmenti%5BMAJR%5D%29+AND+%28incontinentia+pigmenti%5BTIAB%5D%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+1440+days%22%5Bdp%5D>

OMIM

- INCONTINENTIA PIGMENTI
<http://omim.org/entry/308300>

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